



TRAPPC2 gene

trafficking protein particle complex 2

Normal Function

The *TRAPPC2* gene provides instructions for producing the protein sedlin, which is active (expressed) in cells throughout the body. The function of sedlin is unclear. Researchers believe that sedlin is part of a large molecule called the trafficking protein particle (TRAPP) complex, which plays a role in the transport of proteins between various cell compartments (organelles). It is thought that sedlin is located between two organelles, the endoplasmic reticulum and the Golgi apparatus. The endoplasmic reticulum is involved in protein processing and transport, and the Golgi apparatus modifies newly produced proteins. How sedlin participates in the movement of proteins between these two organelles is unknown.

Health Conditions Related to Genetic Changes

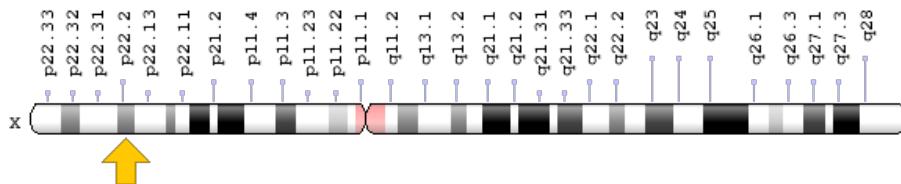
X-linked spondyloepiphyseal dysplasia tarda

At least 40 mutations in the *TRAPPC2* gene have been found to cause X-linked spondyloepiphyseal dysplasia tarda. Most of these mutations delete one or more DNA building blocks (nucleotides) in the *TRAPPC2* gene. All of the mutations result in a nonfunctional sedlin protein. Because sedlin is expressed throughout the body, it is unclear why mutations in the *TRAPPC2* gene affect only bone growth.

Chromosomal Location

Cytogenetic Location: Xp22.2, which is the short (p) arm of the X chromosome at position 22.2

Molecular Location: base pairs 13,712,242 to 13,734,635 on the X chromosome (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- MBP-1 interacting protein-2A
- MIP-2A
- SEDL
- sedlin
- SEDT
- TPPC2_HUMAN
- TRS20
- ZNF547L

Additional Information & Resources

GeneReviews

- X-Linked Spondyloepiphyseal Dysplasia Tarda
<https://www.ncbi.nlm.nih.gov/books/NBK1145>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28TRAPPC2%5BALL%5D%29+OR+%28SEDL%5BTIAB%5D%29+OR+%28sedlin%5BTIAB%5D%29+OR+%28SEDT%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2520+days%22%5Bdp%5D>

OMIM

- TRACKING PROTEIN PARTICLE COMPLEX, SUBUNIT 2
<http://omim.org/entry/300202>

Research Resources

- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=TRAPPC2%5Bgene%5D>
- HGNC Gene Family: Trafficking protein particle complex
<http://www.genenames.org/cgi-bin/genefamilies/set/772>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=23068
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/6399>

- UniProt: TPC2A_HUMAN
<http://www.uniprot.org/uniprot/P0DI81>
- UniProt: TPC2B_HUMAN
<http://www.uniprot.org/uniprot/P0DI82>

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